3125

THE EFFECTS OF TROPICAMIDE 0.5% ON VISUAL FUNCTION

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PURPOSE: To assess the effect of change in pupil diameter induced by Tropicamide 0.5% on Snellen acuity and letter contrast sensitivity in humans.

METHODS: 20 eyes were assessed with the Pelli-Robson CS chart and Snellen chart pre- and post-dilatation (Tropicamide 0.5%), with luminance kept at between 1000 and 1100 lux.

RESULTS: Pupil dilatation was found to have little effect on Snellen acuity: 13 eyes remaining unchanged; six deteriorating by one line and only one eye by two lines. However, letter contrast sensitivity showed a more significant reduction. Only four eyes showed no change; six deteriorated by less than one triplet; seven eyes by between one and two triplets and two eyes by more than two triplets.

CONCLUSION: The fall in contrast senstivity was significantly less than previous studies, using square or sine wave gratings. It was considered that form vision was a major factor in reducing the level of visual disturbance resulting from mydriasis.

3126

PSEUDOXANTHOMA ELASTICUM: A CLINICAL AND GENEALOGICAL STUDY

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Purpose
Pseudoxanthoma Elasticum (PXE), a hereditary systemic disorder of the connective tissue, affects the elastic tissue in the cardiovascular system, the skin and the Bruch membrane in the eye, causing angioid streaks and neovascularisations, ultimately decreasing the visual acuity. A genetic localisation for a gene implicated in PXE is not known. Two autosomal recessive and two autosomal dominant types have been described. We examined 15 patients, of seven nuclear families, living in a genetically isolated area in The Netherlands. Pseudo-dominance within this pedigree could be possible. The aim of the study was to investigate all the family members clinically, and, if possible, to trace common ancestors. Blood samples were drawn in order to perform DNA linkage analysis.
Methods
Clinically a full ophthalmologic, dermatologic and cardiologic investigation was performed. Genealogical investigation made it possible to trace the ancestors back in time to some founder couples, who lived more than 200 years ago. DNA linkage analysis was performed according to standard procedures.
Results
Patients and healthy individuals were identified by the methods described above. The mode of inheritance in this pedigree is most likely autosomal recessive, while sometimes pseudo-dominance occurs. This pedigree proved to be sufficiently informative by computer simulation and statistical analysis. Clinical as well as genealogical data and linkage analysis with candidate loci as fibrillin 5, 15 and 17 will be presented.

3127

Linkage of Autosomal Dominant Iris Hypoplasia to the Rieger Syndrome Locus (4q25)

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Purpose: Iris hypoplasia is an autosomal dominant disorder which is frequently associated with glaucoma. This glaucoma is usually resistant to medical therapy and can lead to blindness.
Methods: A large family of Scandinavian descent with a five generation history of iris hypoplasia was studied. Fifteen individuals were found to have iris hypoplasia, nine of whom had associated glaucoma. In attempt to identify the chromosomal location of the disease causing gene, this family was genotyped with short tandem repeat polymorphisms (STRs) known to map to loci previously associated with glaucoma.
Results: The juvenile glaucoma locus at 1q25 and congenital glaucoma locus on 6p were both excuded. However, significant linkage was demonstrated at the Rieger syndrome locus at 4q25. The highest LOD score was 3.7 (theta=0) and was obtained with marker D4S1616. Three recombination events were observed in affected individuals that together demonstrate that the disease causing gene lies between markers D4S1651 and D4S1611.
Conclusions: These results suggest that autosomal dominant iris hypoplasia and Rieger syndrome may be allelic.

3128

Warburg-Walker syndrome.(Report Of three Cases)
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Purpose: Warburg-Walker syndrome associates agyria hydrocephalus and eye malformations. The aim of this study is to report 3 new cases
Materials and methods: Prenatal diagnosis of these three cases (from two differents families) revealed major anomalies and pathologic examination of brain and eyes were perfomed.
Results: Pathologic examination of the brain showed type II lissencephaly and pathologic examination of the eyes showed retinal dysplasia in all cases.
Discussion: According to Warburg the association of hydrocephalus and agyria with retinal dysplasia constitutes the essential elements for diagnosis. Dobyns required for the diagnosis of the syndrome: type II lissencephaly, cerebellar malformations and congenital muscular dystrophy. The knowlegde of this syndrome emphasizes the need for strict systematic ultrasonographic follow up and genetic advice in families at risk.